Supplementary Figure 1 (Ikeda Y et al)

(a)

Gene	Disease	Number of rare variants: Cases	Number of rare variants: Controls	Samples 1	2	3	4	5	6	7	8	9	10	11	12	13	14
PTEN	Cowden syndrome	0	0														
KLLN		0	0														
SDHB	Hereditary paraganglioma / Pheochromocytoma	0	1														
SDHC		0	2														
SDHD		0	1														
BRCA1	Hereditary breast and ovarian cancer syndrome	1	20									# 1					
BRCA2		2	41										# 3,4				
CDKN2A	Familial atypical multiple mole melanoma syndrome	0	6														
MLH1	Lynch syndrome	0	11														
MSH2		1	4									# 2					
MSH6		0	13														
PMS2		0	8														
APC	Familial adenomatous polyposis	0	40														
MUTYH		0	9														
STK1(LKB1)	Peutz-Jeghers syndrome	0	0														
CDH1	Hereditary diffuse gastric cancer	1	5											# 5			
TP53	Li-Fraumeni syndrome	0	4														
VHL	Von Hippel Li syndrome	0	1														
RB1	Retinoblastoma	0	12														

Mutation

(b)

		Gene	Mutation	Polyphen2 HumDiv	LRT score	MutationTaster	Polyphen2 HumVar	SIFT
Case9	#1	*BRCA1*	L523M					
	#2	*MSH2*	I169V					
Case10	#3	*BRCA2*	N1228D					
	#4	*BRCA2*	R2034C					
Case11	#5	*CDH1*	G693S					

deleterious